



# NEWBORN SCREENING NEWS

The California Newborn Screening Program

Spring 2001

## Repeat Newborn Screening (NBS) Test Required When Initial Specimen is Collected before 12 Hours of Age

Beginning April 2001, as a result of new California State Regulations, newborns screened at less than 12 hours of age, for any reason, will have to be re-screened through the NBS Program. Included in this group are babies whose specimen collection forms have erroneous or missing information, without which the age at collection can not be determined. Licensed perinatal facilities will be required to collect another specimen from these babies on or before the sixth day of age.

The NBS Program has routinely cautioned against early specimen collection. In 1995, the Program issued interim early testing guidelines specifically warning against specimen collection prior to 12 hours of age. While multiple mailings, newsletters, and other attempts to reinforce these recommendations have substantially reduced the number of newborns tested under 12 hours of age, some newborns are still being tested inappropriately. The Program is aware of 3 children with PKU who were not identified through screening because their specimens were collected under 6 hours of age. Because they were not diagnosed and treated soon after birth, these children suffer significant mental retardation.

While licensed perinatal facilities have the primary responsibility to ensure another specimen is collected when an initial specimen is drawn before 12 hours of age, it is the primary care physician's responsibility to verify that every baby in his/her care has been properly tested.

**To minimize the number of newborns requiring a second screen, it is important to adhere to the following NBS Program requirements:**

- All information on the specimen collection form, especially the dates and times of birth and specimen collection, **must** be accurate, complete, and legible.

- Babies **shall** be screened after 12 hours of age, as close to discharge as possible, but no later than the sixth day of age. **The only exceptions are:**

<i>Exception:</i>	<i>Action Required:</i>
Baby is discharged before 12 hours of age	1) Collect specimen prior to discharge 2) Inform parents of need for second specimen after 12 hours but no later than 6 days of age 3) Schedule second collection.
Baby receives a red cell transfusion in the first 12 hours of life	Collect: 1) a specimen prior to transfusion regardless of age 2) a post-transfusion specimen, no sooner than 24 hours after transfusion but no later than 6 days of age when possible.

**The following procedure will be adhered to for specimens collected early or those for which collection time cannot be determined:**

- Newborns with specimens collected at less than 12 hours of age or with insufficient data to calculate age at collection will be reported to the NBS Area Service Centers. The Area Service Center staff will contact the hospitals that collected these specimens to determine actual dates and times of birth and specimen collection and to obtain the scheduled time and date of the repeat.
- All specimens confirmed to have been collected at less than 12 hours of age will have results reported **without** values for the phenylketonuria (PKU) screen and a second specimen will be required.

If there are questions regarding this information, contact your NBS Area Service Center (listed on back page).

# AB 2427 Genetic Diseases and Screening

On September 28, 2000, Governor Gray Davis signed a new bill, AB 2427 (Kuehl), which provides for updating and expanding the Newborn Screening Program in California. This new law took effect on January 1, 2001.

The California Newborn Screening Program, which has been in existence since 1980, currently tests for PKU, Galactosemia, Primary Congenital Hypothyroidism, Sickle Cell Disease and other hemoglobinopathies. AB 2427 requires the Department to establish a new and broader testing program. The new program will include development and evaluation of expanded genetic disease testing utilizing Tandem Mass Spectrometry (MS/MS), which has the potential to test for a wide array of metabolic disorders. Because this technology can process specimens so rapidly, the system can handle the large numbers of samples required in newborn screening. DHS is to provide its own laboratory testing facilities or to contract with qualified laboratories which have the TMS technology.

The current mandatory screening program is supported by newborn screening fees which are deposited into the

Genetic Disease Testing Fund. AB 2427 appropriates \$3.9 million to the Department from the Genetic Disease Testing Fund to fund an expanded screening pilot program. Participation in the pilot program is voluntary. During this period there will be no additional fee for the added tests. At the conclusion of the pilot program, a new fee for the testing will be established by regulations in order to meet the costs of the expanded testing program. These fees will be billed directly to the newborn's parent/guardian, or health care insurance plan.

The Genetic Disease Branch of the DHS has begun the planning process for implementation. A meeting of metabolic and laboratory experts from across the State was held on October 13, 2000 at which time recommendations regarding the specific disorders to be included in the initial pilot phase and the implementation process were discussed. It is anticipated that the pilot program will begin on or shortly after July 1, 2001. A follow-up report is required to be sent to the Legislature on or before January 1, 2002.

## NBS Follow-Up Regions



## Newborn Screening-Area Service Centers (NBS-ASCs)

### CHO

Children's Hospital Oakland (510) 428-3127

### VCH

Valley Children's Hospital (559) 353-6416

### UCLA

UCLA Medical Center (310) 826-4458

### Harbor/UCLA

Harbor/UCLA Medical Center (310) 222-3751

### SDICDSI

San Diego-Imperial Counties

Developmental Services, Inc. (858) 576-2975

### Kaiser N

Kaiser Permanente, Northern CA (510) 596-6192

### Kaiser S

Kaiser Permanente, Southern CA (626) 564-3322

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